

Diagnosis Unknown?



newlife
foundation for disabled children

Action for disabled & terminally ill children in th UK

Diagnosis Unknown?

“Not knowing what’s wrong, why it happened, what its called - its just terrible” **Mother**

“They say knowledge is power and because I don’t know why or what my son’s condition is called I feel so powerless to help him - what if I could be doing more, if only I knew” **Father**

“It is believed that thousands of parents and their children remain without ever knowing what condition affects their child” **Newlife Foundation**

Index

- “Why can’t the doctors tell us why our child is like this?”
- “How can a Doctor help to find out a name for my child’s condition?”
- “My child was tested when he was young but they didn’t know what was wrong then. Could they tell now there are new tests?”
- “Does getting a diagnosis make a difference/matter?”
- “I always thought my child was this way because of something I did – I have always felt guilty about this – could having a diagnosis help?”
- “How do I go about pursuing a diagnosis?”
- “What can I do to help the Doctors find a diagnosis?”
- “What I know or see”
- “When people ask me what he/she has I never know what to say – can you suggest something?”
- “Will my child be negatively ‘labelled’ if I find a specific condition name for the disorder?”
- “Will knowing a specific disorder name change how I see/appreciate/treat my child?”

Diagnosis Unknown?

“Why can’t the Doctors tell us why our child is like this?”

Be assured that Doctors do generally want to help you find a named diagnosis for your child. However some conditions are harder to diagnose than others. It is also sometimes the case that having a named diagnosis would not alter the treatment, therapy or outcome for a child and so sometimes the name of the condition is not as strenuously pursued as perhaps a parent may desire.

Sadly, it is likely that some conditions will never have a specifically named diagnosis. This is because each individual person/child is a truly unique arrangement of genes gained from his/her parents and their ancestors and, in addition to this, new arrangements of genes can occur with each conception, so finding a diagnosis can be very difficult. This is not because no one cares but because, at the present time, thousands of conditions remain un-named and un-researched. However, more genes are being identified each month and, therefore, medical research could well challenge this situation in the years ahead. A Doctor can only use the tools he has available to carry out tests to try to identify a condition. Because of the number of conditions which can be inborn, sometimes a Doctor may never have seen a case of a particular condition so he/she may need to make referrals to other specialists, such as a Geneticist or a specialist Paediatrician, to help make a diagnosis.

“How can a Doctor help to find out a name for my child’s condition?”

There are two basic ways that conditions are diagnosed, these can be used alone and in conjunction with one another :

- Clinical tests – physical test using body samples or images, etc.
- Clinical feature recognition - assessment of symptoms gained from physical examination or assessments

While medical research has made major advances, much more investment is needed to increase the number of conditions which can be tested for. However, blood and other samples can be used in the laboratory to detect abnormalities.

Diagnosis Unknown?

Commonly, these tests look for abnormal cells, proteins, etc. but over the last few years the role of gene and chromosome tests has become a major method of confirming diagnosis. Despite this, not all conditions can be detected even using the most up to date techniques.

“My child was tested when he was young but they didn’t know what was wrong then. Could they tell now there are new tests?”

Many tests have developed greatly over the past few years, especially genetic and chromosome testing. Therefore, those children who have had tests, which were carried out over 10/15 years ago could be tested again using more recent techniques.

“Does getting a diagnosis make a difference/matter?”

In some cases where treatment, therapy or care are good, in hand, and all that can be done is happening, pursuing a confirmed diagnosis may not be thought to be necessary. However, as it is now known that many conditions are genetic in nature, there could be a long-term benefit to the child/person affected, in terms of their own reproductive options, and, so that their brother, sister, or close family members can make informed decisions about their own reproduction.

“I always thought my child was this way because of something I did – I have always felt guilty about this – could having a diagnosis help?”

Almost every parent of a child with an inborn condition asks, “why?”. It’s perfectly natural to be concerned about why a condition occurred and it’s natural to want to find out some answers. Pursuing a diagnosis may or may not achieve a specifically named diagnosis but a specialist would probably be able to clear up this concern for you.

Diagnosis Unknown?

Many parents believe they have contributed to their child's condition but this is usually quite untrue and in addition to these worries there are also all sorts of myths and folk lore/old wives tales about why inborn conditions occur.

The queries shown below are all questions received from real parents who have real concerns about what could have caused their child's condition:

"I was on a diet before I conceived and I didn't know I was pregnant for a while so I was eating well in the first few weeks"

"My husband worked in an industry where there have been rumours that health is affected – could this have caused this condition?"

"I unknowingly used an insecticide while pregnant – could this have caused the condition?"

"My grandparents were 1st cousins"

"My child was conceived out of wedlock"

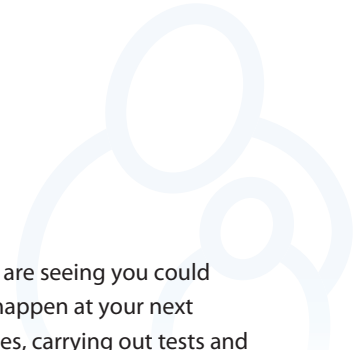
"I had a cold during pregnancy"

"My husband smoked while I was pregnant"

"I had two sherries at Christmas while pregnant"

From practical experiences which concern parents, about why the condition occurred, to cultural and religious perceptions, which can have equally powerful influences, all can cause unnecessary worrying and guilt by parents. Most parents find that by openly asking about these issues and raising their concerns, Doctors can clear up these matters and help overcome negative feelings about these worries and concerns. In pursuing a diagnosis, answers to these sorts of questions can also be responded to. Don't worry – Doctors hear questions like this all the time – they won't be shocked or surprised and will be sensitive to your queries if you are clear about how much the issue concerns you.

Diagnosis Unknown?



“How do I go about pursuing a diagnosis?”

If your child is under 16 and you have a Paediatrician you are seeing you could ask the Paediatrician to review the diagnosis. This could happen at your next routine appointment. He or she can pursue this themselves, carrying out tests and assessments, or they could refer to other specialists including a Geneticist. If under 16, but you do not see a Paediatrician or over 16, and not seeing a Paediatrician, you could ask your GP for a referral to a Paediatrician as you want the diagnosis reviewed.

You should explain to the GP or the Paediatrician why you want the diagnosis reviewed. Explain that you want to understand whether or not the condition could be genetic and, as new tests are now available which were not possible when the child was much younger, you feel it is time to pursue this. Explain that you are concerned about why the condition has occurred and be honest and explain that this troubles you and you feel you need some answers. You could also explain that you have concerns for the child’s future reproduction options and for the reproductive options of brothers and sisters, etc., in case it is a genetic condition. You can explain that you feel that knowing what the condition is would help you manage the situation better and feel more able to cope, not only with the day to day practicalities but in planning care for the future.

Given such well explained reasons, most Doctors will feel able to make a referral in pursuit of a diagnosis.

“What can I do to help the Doctors find a diagnosis?”

Before being referred to a specialist to pursue a diagnosis there are things that you could do to help:

Family History

Look and note brief details of your family history and briefly record any instances of repeated miscarriages, stillbirths, infant deaths, long-term conditions. Look at parents, maternal and paternal grandparents, brothers and sisters of the child, other children of you or your partner, the brothers and sisters of you and your partner.

Diagnosis Unknown?

Write down any experience/conditions and explain these in relationship to your child (not the relationship to you/your partner). Remember, some conditions are basically late onset conditions in aged relatives so these do not need to be noted, for example, a grandfather may have died of a heart condition which only became a problem when he was aged. However, if the person suffered from a heart condition from being a child, this is worth noting. It is probably better to include all information so the Doctor can discount it. Always be honest about genetic relationships, e.g. if the child is adopted, or if the family history of one parent is unclear, this can occur because of adoption, etc. This information can have a significant effect on the ability to diagnose or not.

What you know/see

Before the appointment you could also write down what you see/believe are the problems or symptoms in the form of a list, copy it and give it to the Doctor/s concerned. This should be written as an aid to the appointment and to help you remember everything, but do write down significant issues. He/she does not need to know about every cold or measles.

Try to break these down into sections such as:

Organs and digestion - issues with digestion, urination, bowels, heart, liver, kidneys, feeding, genital problems, etc.

Skin/hair/nails and teeth - issue with skin tissue, healing, bruising, hair growth and loss, teeth development and loss, etc.

Joints/muscles - mobility, stiffness, pains, stability, injury issues, co-ordination, restrictions, stamina on exercise, etc.

Senses - hearing, sight, touch, taste, smell – lack of , excessive, poor, restrictions, etc.

Pain and sporadic incidents - if pain follows activity or incidents, time of day, etc., or if the child appears to lose consciousness or has episodes of seizures or periods of absence, etc.

Diagnosis Unknown?

Mental and emotional development, behaviour and communication – perhaps the hardest of areas to describe, as most of this is relevant to the age of the child at the time. Try to centre on learning ability, milestones achieved or not, speech and ability to understand, ability to make decisions, ability to avoid injury or be safe. Excessive dependence, not acting as one would expect for age. Responses to others, outbursts. Whilst telling the Doctor how these behaviours make you feel may help you, it may not be the best way to help him/her see the practical problems and symptoms.

Other symptoms – these may include things like unusual sweats, smells, perspiration, soreness, tremors, habits such as repeated blinking or picking at things, obsessions, repeated infections, serious injuries and infections or surgery, etc.

“When people ask me what he/she has, I never know what to say – can you suggest something?”.

It is so difficult not to have a specific named condition but many of these disorders are apparent or exist from birth (i.e. not caused by illness/infection/accident in childhood, etc.) Therefore many families find it easiest to explain, in shorthand terms, by saying “he/she has an MCA.” The term MCA is routinely used by the medical world to describe a non-specific Multiple Congenital Anomaly, otherwise known as an inborn condition. For this reason many parents will explain their child’s condition using a short phrase such as : “Johnny had an MCA with epilepsy and speech problems.” Or they may say “Sarah has an MCA with hearing problems and autistic traits”. This helps because it is short and quick to say and sums up the main points. There may be other symptoms but most people just need a shorthand description of the condition comprising the main points.

You don’t need to wait for a Doctor to say this, you, yourself, can start to use this phrase, just work out a short form of words which starts with (child’s name) has an MCA with _____ and _____.

Any Doctor you meet will recognise the terminology and you can simply add to the statement by saying . . . “ the specific condition is currently un-diagnosed”.

Diagnosis Unknown?

“Will my child be negatively ‘labelled’ if I find a specific condition name for the disorder?”.

No, this concern was a reality maybe 30 years ago when the understanding of inborn and other conditions, illnesses and disabilities was poor. But no child these days would be automatically assessed for services or ability levels just on the basis of a named diagnosis. This is because standards, understanding and appreciation of the whole, and specialist needs and abilities of the individual person have greatly improved since then. These days, all care plans, education and treatment is solidly based on detailed individual assessments. Even children with the same named condition can have dramatically different care or support as the individual traits or features of the condition can be unique to that child.

“Will knowing a specific disorder name change how I see/ appreciate/treat my child?”

Most parents say that knowing a named condition helps them. I needed to know, what?, and why?, and, when I found out, I was able to use the information where it was useful, but most of the time I just think of Susie as Susie, not Susie with this syndrome. This is the most common experience of families. They felt the benefit from a diagnosis, but once they have learnt about it and understood and know when and where to use the proper name – they just put it aside and treat the child as they did before, but perhaps with greater insight into why some things are the way they are, and most families report that this helps them cope better.





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